



Beacon Carrier Screening – 466 genes This analysis was run using the Beacon Carrier Screening gene list. 466 genes were tested with 99.6% of targets sequenced at >20x coverage. For merged couples reports, X-linked genes are evaluated in the female partner only.

ABCA3, ABCA4, ABCB11, ABCC8, ABCD1, ABCD4, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACP5, ACSF3, ADA, ADAMTS2, ADGRG1, ADK, AFF2, AGA, AGL, AGPS, AGXT, AHCY, AH11, AIPL1, AIRE, ALDH3A2, ALDH4A1, ALDOB, ALG6, ALMS1, ALPL, AMT, ANO10, APIS2, AQP2, ARG1, ARL13B, ARSA, ARSB, ARSE, ARX, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7A, ATP7B, ATRX, BBS1, BBS10, BBS12, BBS2, BCHE, BCKDHA, BCKDHB, BCS1L, BLM, BRWD3, BSND, BTD, CAPN3, CASQ2, CBS, CC2D2A, CCDC103, CCDC151, CCDC39, CCDC88C, CD40LG, CDH23, CEP290, CERKL, CFTR, CHM, CHRNE, CHNG, CHST6, CIITA, CLCN1, CLCNKB, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, COL27A1, COL4A3, COL4A4, COL4A5, COL6A2, COL7A1, COLQ, CPS1, CPT1A, CPT2, CRADD, CRB1, CRYL1, CTNS, CTSK, CUL4B, CYBA, CYBB, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, DBT, DCLRE1C, DCX, DDX11, DHCR7, DHDDS, DLD, DLG3, DMD, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DPYD, DUOX2, DUOXA2, DYNC2H1, DYSF, EDA, EIF2AK3, EIF2B5, ELP1, EMD, ERCC2, ERCC3, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EYS, F11, F5, F8, F9, FAH, FAM161A, FANCA, FANCC, FANCG, FGD1, FH, FKRP, FKTN, FMO3, FMRI, FTCD, FTSJ1, FXN, G6PC, G6PD, GAA, GALT, GALE, GALK1, GALNS, GALT, GAMT, GBA, GBE1, GCDH, GDAPI, GFM1, GJB1, GJB2, GJB6, GLA, GLB1, GLDC, GLE1, GNE, GNPTAB, GNPTG, GNRHR, GNS, GPIBA, GP9, GPR143, GRHRP, GRIP1, GUCY2D, GUSB, HADHA, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGD, HGSNAT, HJV, HLCS, HMGCL, HOGA1, HPS1, HPS3, HSD17B4, HSD3B2, HYAL1, HYLS1, IDH3B, IDS, IDUA, IL1RAPL1, IL2RG, IVD, IYD, JAK3, KCNJ1, KCNJ11, KDM5C, L1CAM, LAMA2, LAMA3, LAMB3, LAMC2, LCA5, LDLRAP1, LHX3, LIFR, LIPA, LMBRD1, LOXHD1, LPL, LRP2, LRPPRC, LYST, MAN2B1, MCCC1, MCCC2, MCEE, MCOLN1, MCPH1, MECP, MED17, MEFV, MESP2, MFSD8, MID1, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MPI, MPL, MPV17, MTHFR, MTM1, MTMR2, MTRR, MTPP, MUT, MVK, MYO15A, MYO7A, NAGA, NAGLU, NAGS, NBN, NDP, NDRG1, NDUFAF5, NDUFS6, NEB, NHEJ1, NPC1, NPC2, NPHP1, NPHS1, NPHS2, NROB1, NR2E3, NTRK1, OAT, OCA2, OCRL, OPA3, OPHN1, OTC, OTOF, P3H1, PAH, PAK3, PANK2, PC, PCBD1, PCCA, PCCB, PCDH15, PDE6A, PDHA1, PDHB, PEX1, PEX10, PEX12, PEX2, PEX6, PEX7, PFKM, PGK1, PHF8, PHGDH, PKHD1, PLA2G6, PLOD1, PLP1, PMM2, POLG, POLR1C, POMGNT1, POMGNT2, POMT1, POMT2, POU3F4, PPT1, PQBP1, PRF1, PROP1, PRPS1, PSAP, PTS, PUS1, PYGM, QDPR, RAB23, RAG1, RAG2, RAPSN, RARS2, RAX, RDH12, RMRP, RNASEH2B, RP2, RPE65, RPGR, RPGRIP1L, RS1, RTEL1, SACS, SAMD9, SAMHD1, SCO2, SEPSECS, SERPINA1, SGCA, SGCB, SGCD, SGCG, SGSH, SH3TC2, SLC12A3, SLC12A6, SLC16A2, SLC17A5, SLC19A3, SLC1A4, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC26A2, SLC26A3, SLC26A4, SLC2A2, SLC35A3, SLC37A4, SLC38A8, SLC39A4, SLC46A1, SLC4A11, SLC5A5, SLC6A19, SLC6A8, SLC7A7, SMARCAL1, SMN1, SMPD1, SPG11, SPG7, SPINK5, STAR, SUMF1, SURF1, SYN1, SZT2, TAT, TBCD, TCIRG1, TCTN2, TECPR2, TF, TFR2, TG, TGM1, TH, THOC2, TK2, TKT, TMC1, TMEM216, TMEM67, TNXB, TPO, TPP1, TRDN, TRIM32, TRMU, TSFM, TSHB, TSPEAR, TTC37, TTPA, TYMP, TYR, UGT1A1, UPB1, UPF3B, USH1C, USH1G, USH2A, VPS13A, VPS13B, VPS45, VPS53, VRK1, VSX2, WAS, WHRN, WNT10A, WWOX, XPA, XPC, ZDHHC9, ZFYVE26, ZNF711